



Childhood absence epilepsy

Childhood absence epilepsy is a condition characterized by recurrent seizures (epilepsy). This condition begins in childhood, usually between ages 3 and 8. Affected children have absence seizures (also known as petit mal seizures), which are brief episodes of impaired consciousness that look like staring spells. During seizures, children are not aware of and do not respond to people or activities around them. The seizures usually last several seconds and they occur often, up to 200 times each day.

Some affected individuals have febrile seizures before they develop childhood absence epilepsy. Febrile seizures are involuntary muscle contractions (convulsions) brought on by a high body temperature (fever).

In most people with childhood absence epilepsy, the absence seizures disappear in adolescence. However, some affected individuals continue to have absence seizures into adulthood, or they may develop generalized tonic-clonic seizures, which cause muscle rigidity, convulsions, and loss of consciousness, or myoclonic seizures, which are characterized by rapid, uncontrolled muscle jerks.

Frequency

Childhood absence epilepsy affects 2 to 8 in 100,000 children under age 15 each year. The condition is more common in girls than in boys.

Causes

The genetics of childhood absence epilepsy are complex and not completely understood. It is thought that multiple genetic changes or a combination of genetic and environmental factors contribute to development of the condition. Most genetic changes associated with childhood absence epilepsy are rare, having been found in only a small number of affected individuals. Each genetic change appears to play a role in some populations but not others.

Several genes associated with childhood absence epilepsy provide instructions for making pieces (subunits) of the GABA_A receptor protein. The GABA_A receptor acts as a channel that allows negatively charged chlorine atoms (chloride ions) to cross the cell membrane. The influx of chloride ions in nerve cells (neurons) in developed brains creates an environment that blocks (inhibits) signaling between neurons and prevents the brain from being overloaded with too many signals. Mutations in GABA_A receptor subunit genes lead to production of altered subunit proteins that cannot form functional receptors, so fewer GABA_A receptors are available. As a result, neurons become overloaded with signals. Researchers believe that the overstimulation of certain neurons in the brain triggers the abnormal brain activity associated with seizures.

Problems with another type of ion channel, called a calcium channel, are also associated with childhood absence epilepsy. Calcium channels transport positively charged calcium atoms (calcium ions) into cells. These channels help control the release of neurotransmitters, which are chemicals that relay signals from one neuron to another. Mutations that result in overactive calcium channels cause certain neurons to become overstimulated, triggering seizures.

Mutations in other genes that do not provide instructions for making ion channels have also been associated with childhood absence epilepsy. It is not clear how these changes are involved in the development of absence seizures.

Inheritance Pattern

Because childhood absence epilepsy appears to be a complex disease without a single genetic cause, it does not have a straightforward pattern of inheritance. When associated with mutations in GABA_A receptor or calcium channel genes, it seems to follow an autosomal dominant inheritance pattern, which means one copy of the altered gene in each cell is sufficient to increase the likelihood of the disorder. Some people who have the altered gene never develop the condition, a situation known as reduced penetrance.

Other Names for This Condition

- absence epilepsy, childhood
- petit mal epilepsy
- pykno-epilepsy
- pyknolepsy

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Epilepsy, childhood absence 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838604/>
- Genetic Testing Registry: Epilepsy, childhood absence 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1843244/>
- Genetic Testing Registry: Epilepsy, childhood absence 4
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970160/>
- Genetic Testing Registry: Epilepsy, childhood absence 5
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2677087/>

- Genetic Testing Registry: Epilepsy, childhood absence 6
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2749872/>
- Genetic Testing Registry: Epilepsy, childhood absence, JRK related
<https://www.ncbi.nlm.nih.gov/gtr/conditions/CN240376/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22childhood+absence+epilepsy%22>

Other Diagnosis and Management Resources

- Children's Hospital of Pittsburgh
<https://www.chp.edu/our-services/brain/neurology/epilepsy/types/syndromes/childhood-absence>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Absence Seizure
<https://medlineplus.gov/ency/article/000696.htm>
- Encyclopedia: Febrile Seizure
<https://medlineplus.gov/ency/article/000980.htm>
- Encyclopedia: Generalized Tonic-Clonic Seizure
<https://medlineplus.gov/ency/article/000695.htm>
- Health Topic: Epilepsy
<https://medlineplus.gov/epilepsy.html>
- Health Topic: Seizures
<https://medlineplus.gov/seizures.html>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: The Epilepsies and Seizures: Hope Through Research
<https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Hope-Through-Research/Epilepsies-and-Seizures-Hope-Through>

Educational Resources

- Epilepsy Foundation New England: Absence Seizures
<https://epilepsynewengland.org/knowledge-center/types-of-seizures/generalized-non-motor-absence-seizures>
- International League Against Epilepsy
<https://www.epilepsydiagnosis.org/syndrome/cae-overview.html>

- KidsHealth from Nemours
<https://kidshealth.org/en/parents/childhood-absence-epilepsy.html>
- MalaCards: childhood absence epilepsy
https://www.malacards.org/card/childhood_absence_epilepsy
- Orphanet: Childhood absence epilepsy
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64280
- The Hospital for Sick Children (Canada)
<https://www.aboutkidshealth.ca/Article?contentid=2076&language=English>

Patient Support and Advocacy Resources

- American Epilepsy Society
<https://www.aesnet.org/>
- Epilepsy Foundation
<https://www.epilepsy.com/learn/professionals/about-epilepsy-seizures/overview-epilepsy-syndromes/childhood-absence-pyknolepsy>
- Epilepsy Society (UK)
<https://www.epilepsysociety.org.uk/>
- Medical Home Portal
<https://www.medicalhomeportal.org/diagnoses-and-conditions/childhood-absence-epilepsy>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Epilepsy,+Absence%5BMAJR%5D%29+AND+%28childhood+absence+epilepsy%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 1
<http://omim.org/entry/600131>
- EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 5
<http://omim.org/entry/612269>
- EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 6
<http://omim.org/entry/611942>
- FEBRILE SEIZURES, FAMILIAL, 8
<http://omim.org/entry/607681>

Medical Genetics Database from MedGen

- Childhood absence epilepsy
<https://www.ncbi.nlm.nih.gov/medgen/924120>
- Epilepsy, childhood absence 1
<https://www.ncbi.nlm.nih.gov/medgen/325057>
- Epilepsy, childhood absence, JRK related
<https://www.ncbi.nlm.nih.gov/medgen/910256>

Sources for This Summary

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Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/27367160>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5426359/>
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Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22206818>

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